



What is 22q11.2 Deletion Syndrome?

22q11.2 deletion syndrome is a genetic condition that affects learning, health, and physical traits. It occurs in males and females of all racial and ethnic backgrounds. 22q11.2 deletion syndrome occurs in about 1 in 4,000 to 6,000 people. It is sometimes known as Velocardiofacial or DiGeorge syndrome.

Michigan Resources & Support

Children's Special Health Care Services

Family Phone Line
Toll-free: 1-800-359-3722
E-mail: ppp@michigan.gov
www.michigan.gov/cshcs

Early On[®] Michigan

Toll-free: 1-800-EARLY ON
www.1800earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection

www.migeneticsconnection.org

National Resources & Support

Chromosome 22 Central

www.nt.net/~a815/chr22.htm

International 22q11.2 Deletion Syndrome Foundation

Toll-free: 1-877-739-1849
www.22q.org/

Velo-Cardio-Facial Syndrome Educational Foundation

Toll-free: 1-866-VCFSEF5
www.vcfsef.org

Family Village

www.familyvillage.wisc.edu

GeneReviews

www.geneclinics.org/profiles/22q11deletion

Genetic and Rare Diseases Information Center

Toll-free: 1-888-205-2311
E-mail: GARDinfo@nih.gov

Genetics Home Reference

www.ghr.nlm.nih.gov/condition=22q112deletionsyndrome

How may the syndrome affect my child?

Learning: There is a range of abilities. Some children with 22q11.2 deletion may have normal learning, while others have a developmental delay, learning disability or severe lifelong learning problems.

Behavior: Mental (psychiatric) illnesses including depression, bipolar disorder and schizophrenia have been reported in some people with 22q11.2 deletion syndrome.

Physical: Children with 22q11.2 deletion syndrome may have certain facial features, such as a prominent bridge of the nose and narrow eyes,

often noticed only by a medical professional. Short stature is also a common trait.

Medical: Heart defects are very common. They can be mild or severe enough to need surgery. Many children have palate abnormalities. These include an opening in the roof of the mouth (cleft palate) and a change in the way the throat closes (velopharyngeal incompetence). Other medical problems can include kidney abnormalities, problems with the immune system and low calcium levels.

How does the syndrome occur?

22q11.2 deletion syndrome is caused by a change in the #22 chromosome. A very small piece of the chromosome is missing (deleted), including some of the genes within it. The child is often the first and only family member affected, but sometimes the deleted chromosome is passed down from a parent. A person with the 22q11.2 deletion has a 1 in 2 (50%) chance of passing it on to each of his or her children. Genetic counseling is recommended for parents to learn about the genetic cause of the syndrome in their family, and possible health risks for other children.

How is the syndrome treated?

22q11.2 deletion syndrome cannot be cured, but many symptoms can be treated. Surgery may be needed to repair a heart defect or cleft palate. The immune system should be tested, and the kidneys should be checked by ultrasound in infancy. Calcium supplements may be needed. Infants and toddlers (birth to 3 years) should be connected with *Early On*[®] Michigan as soon as possible. If there are concerns about learning, speech, or behavior in a child over 3 years of age, a referral for special education services should be made. Other therapies or treatments may be available for problems as they arise. Children with 22q11.2 deletion syndrome and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov

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What is Beckwith-Wiedemann Syndrome?

Beckwith-Wiedemann syndrome (BWS) is a genetic condition that affects growth. It occurs in males and females of all ethnic and racial backgrounds. It causes rapid or uneven growth, low blood sugar and other physical features. BWS occurs in about 1 in 13,000 people.

Michigan Resources & Support

Children's Special Health Care Services

Family Phone Line
Toll-free: 1-800-359-3722
E-mail: ppp@michigan.gov
www.michigan.gov/cshcs

Early On[®] Michigan

Toll-free: 1-800-EARLY ON
www.1800earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection

www.migeneticsconnection.org

National Resources & Support

Beckwith-Wiedemann Support Network

www.beckwith-wiedemann.org/

Beckwith-Wiedemann Syndrome Children's Foundation

Phone: 425-338-4610
www.beckwith-wiedemannsyndrome.org

Beckwith-Wiedemann Syndrome Family Forum

www.geocities.com/beckwith_wiedemann

BWS Registry

E-mail: bwsregistry@kids.wustl.edu

Family Village

www.familyvillage.wisc.edu

GeneReviews

www.geneclinics.org/profiles/bws

Genetic and Rare Diseases Information Center

Toll-free: 1-888-205-2311
E-mail: GARDinfo@nih.gov

How may BWS affect my child?

Learning: Most children with BWS have normal learning unless medical complications result in delayed development.

Physical: An opening in the abdomen is common. This may consist of a bulging near the belly button (hernia) or a large protrusion that contains some of the intestines (omphalocele). Kidney abnormalities, cleft palate and creases or pits in the earlobe are also common. Large body size (macrosomia) and enlarged tongue (macroglossia) are usually present at birth or develop in early

childhood. Children with BWS tend to grow rapidly and are big for their age. Uneven (asymmetric) overgrowth of parts of the body may also occur.

Medical: There is an increased risk (~7.5%) for certain types of tumors. The risk appears to be the greatest during the first eight years of life. The tumors can be benign or cancerous. Low blood sugar (hypoglycemia) is common in newborns. If untreated, the child's development may be affected. Thyroid levels may be low, while cholesterol is often high.

How does BWS occur?

There are different ways BWS can occur. Most often, it is caused by changes in genes located on the #11 chromosome. The child with BWS is usually the first and only family member affected, but sometimes the condition is passed down from a parent. Genetic counseling is recommended for parents to learn about the genetic cause of BWS in their family, and possible health risks for other children.

How is BWS treated?

BWS cannot be cured, but many symptoms can be treated. The effects of BWS are seen mainly in childhood. Complications occur less often in adults. Soon after birth, low blood sugar must be treated. Surgery may be needed to repair an abdominal wall or other birth defect. Frequent screening (about every 3 months) using blood tests and ultrasound is needed to check for tumors during the first 8 years. When a large tongue is present, evaluation by a craniofacial team helps to ensure proper management. Other treatments may be needed for health problems as they arise. Infants and toddlers (birth to 3 years) should be connected with *Early On*[®] Michigan if there are concerns about learning, speech, or behavior; while children over 3 years of age should be referred for special education services if concerns arise. Children with BWS and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov

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What is Down Syndrome?

Down syndrome is a genetic condition that occurs in males and females of all racial and ethnic backgrounds. It causes different types of birth defects, as well as developmental disabilities. Down syndrome occurs in about 1 in 800 people.

Michigan Resources & Support

Down Syndrome Support Groups

www.migeneticsconnection.org

click on "support group directory"

Children's Special Health Care Services

Family Phone Line

Toll-free: 1-800-359-3722

E-mail: ppp@michigan.gov

www.michigan.gov/cshcs

Early On[®] Michigan

Toll-free: 1-800-EARLY ON

www.1800earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator

Toll-free: 1-866-852-1247

E-mail: BDRfollowup@michigan.gov

Project PERFORM

www.wash.k12.mi.us/perform/

The Arc Michigan- Family Information Exchange

www.arcmi.org/FIE.htm

National Resources & Support

National Down Syndrome Congress

Toll-free: 1-800-232-6372

www.ndsccenter.org

National Down Syndrome Society

Toll-free: 1-800-221-4602

www.ndss.org

Family Village

www.familyvillage.wisc.edu

Genetics Home Reference

www.ghr.nlm.nih.gov/condition=downsyndrome

KidsHealth.org

www.kidshealth.org/parent/medical/genetic/down_syndrome.html

March of Dimes

www.marchofdimes.com

click on Birth Defects

How may Down syndrome affect my child?

Learning: There is wide variation in the mental abilities of children with Down syndrome. Most have developmental delays. They usually learn at a slower pace, but do not lose skills once they are acquired. They may also have trouble with judgment and reasoning. The degree of mental impairment is usually in the mild to moderate range.

Behavior: Emotional problems such as behavior issues or depression may occur beginning in childhood.

Physical: Common facial features include upward slanting eyes with epicanthal folds (skin over the inside

corner of the eye), a small mouth, and a flat nasal bridge. Children with Down syndrome are often shorter than average, and prone to extra weight gain. Babies may seem "floppy" due to low muscle tone (hypotonia).

Medical: About 1 in 2 children has a heart defect, which can range from minor to very serious. Other common health concerns include vision or hearing problems, thyroid disease, and unstable bones in the neck (atlantoaxial instability). Health problems such as seizures, diabetes, low resistance to infections, intestinal obstruction, and leukemia also affect some people with Down syndrome.

How does Down syndrome occur?

Down syndrome is caused by an extra copy of the #21 chromosome. The child is usually the first and only family member affected, but sometimes there is a family history. In a few families, the extra chromosome is passed down from a parent. The chance of having a baby with Down syndrome increases with a woman's age. Genetic counseling is recommended for parents to learn more about the cause of Down syndrome in their family, and possible risks for other children or relatives.

How is Down syndrome treated?

Down syndrome cannot be cured, but some symptoms can be treated. All children with Down syndrome should be checked for heart defects to find out if medication or surgery is needed. Infants and toddlers (birth to 3 years) should be connected with *Early On*[®] Michigan as soon as possible; while children over 3 years of age should be referred for special education services to address developmental concerns. Physical and occupational therapies may be helpful. Thyroid levels should be checked, and vision/hearing should be screened on a regular basis. Other treatments may be needed for health problems as they arise. Children with Down syndrome and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov

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What is Fragile X Syndrome?

Fragile X syndrome is a genetic condition that occurs in people of all racial and ethnic backgrounds. It is more common in males, but also affects females. Fragile X syndrome is a developmental disorder that affects learning and behavior. It occurs in about 1 in 2,000 people.

Michigan Resources & Support

Fragile X Association of Michigan

Phone: 313-381-2834

www.fragilex.org/html/michigan.htm

Children's Special Health Care Services

Family Phone Line

Toll-free: 1-800-359-3722

E-mail: ppp@michigan.gov

www.michigan.gov/cshcs

Early On[®] Michigan

Toll-free: 1-800-EARLY ON

www.1800earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator

Toll-free: 1-866-852-1247

E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection

www.migeneticsconnection.org

National Resources & Support

National Fragile X Foundation

Toll-free: 1-800-688-8765

www.fragilex.org/

The Fragile X Research Foundation (FRAXA)

Phone: 978-462-1866

www.fraxa.org/

Family Village

www.familyvillage.wisc

GeneReviews

www.geneclinics.org/profiles/fragilex/

Genetics Home Reference

www.ghr.nlm.nih.gov/condition=fragilex/syndrome

National Institute of Child Health and Human Development

www.nichd.nih.gov/publications/pubs/fragileX/index.htm

How may Fragile X syndrome affect my child?

Learning: Early motor skills, speech, and language development are commonly delayed in childhood. People with fragile X syndrome usually have a learning disability. The degree may range from mild impairment to severe mental retardation. Females are often less severely affected than males.

Behavior: Behavioral problems are common in both males and females. These may include unusual hand movements, hyperactivity, anxiety, autism spectrum disorder, and other behaviors.

Physical: Certain facial features may be noticed by a medical professional. These

include a long face, prominent forehead and chin, and large ears. Head size may be larger than average. The joints may be especially flexible. After puberty, large testes are common in males.

Medical: Lazy eye, curvature of the spine (scoliosis), seizures (epilepsy) or heart murmurs occur in some people with Fragile X. Women who carry a certain genetic change may have early menopause. Older adults who are carriers may develop a condition that causes tremors and affects balance and memory, similar to Parkinson's disease.

How does Fragile X syndrome occur?

Fragile X syndrome is caused by a change (mutation) in a gene on the "X" chromosome. Fragile X syndrome may run in a family. Women who carry the mutation, or a smaller change called a "premutation", are more likely to have affected children. Men who carry the premutation are not expected to have affected children, but their grandchildren (daughter's children) may have Fragile X syndrome. The genetics of Fragile X syndrome is complex. Genetic counseling is recommended for parents to learn more about Fragile X syndrome in their family, and possible risks for their children.

How is Fragile X syndrome treated?

Fragile X syndrome cannot be cured, but some symptoms can be treated. Infants and toddlers (birth to 3 years) should be connected with *Early On*[®] Michigan as soon as possible. If there are concerns about learning, speech, or behavior in a child over 3 years of age, a referral for special education services should be made. Children often benefit from physical and occupational therapy, sensory integration, and tools to assist with communication. Other therapies or treatments may be needed for behavioral or health problems as they arise. Children with Fragile X syndrome and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov

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What is Klinefelter Syndrome?

Klinefelter syndrome is a genetic condition that occurs in males of all racial and ethnic backgrounds. It affects sexual development and may lead to learning disabilities. Klinefelter syndrome occurs in about 1 in 500 to 1 in 1,000 boys.

Michigan Resources & Support

Children's Special Health Care Services

Family Phone Line
Toll-free: 1-800-359-3722
E-mail: ppp@michigan.gov
www.michigan.gov/cshcs

Early On[®] Michigan

Toll-free: 1-800-EARLY ON
www.1800earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: BDRfollowup@michigan.gov

National Resources & Support

American Association for Klinefelter Syndrome Information & Support

Toll-free: 1-888-466-KSIS
www.aaksis.org

Klinefelter Syndrome & Associates

Toll-free: 1-888-999-9428
www.genetic.org/ks

Family Village

www.familyvillage.wisc.edu

Genetics Home Reference

www.ghr.nlm.nih.gov/condition=klinefeltersyndrome

National Institute of Child Health and Human Development

www.nichd.nih.gov/publications/pubs/klinefelter.htm

How may Klinefelter syndrome affect my child?

Learning: Most boys with Klinefelter syndrome have normal intelligence. They may have developmental delays in some motor skills like learning to walk. They may also have learning disabilities, especially with speech, language, and reading.

Behavior: While personality and behavior vary, many children are described as being quiet, passive or shy. They may be immature for their age, and have difficulty with social skills. As boys with Klinefelter syndrome grow up, they may have concerns with body image or self-esteem. Emotional problems can include anxiety or depression.

Physical: The features of Klinefelter syndrome are often so mild that a person may not be diagnosed until there are concerns about sexual development or fertility. During puberty, increased body fat around the torso and breast enlargement (gynecomastia) may occur. Facial and body hair is often sparse without hormone therapy and the testes are usually small. Adult height may be taller than other family members.

Medical: There is an increased risk for health problems such as diabetes, thyroid disease, weak bones (osteoporosis), and breast cancer. Most men have normal sexual function, but are infertile.

How does Klinefelter syndrome occur?

Klinefelter syndrome is caused by an extra copy of an "X" chromosome. Usually males have one Y chromosome and one X chromosome in all of the body's cells. Boys with Klinefelter syndrome have a Y and two X chromosomes. The child is usually the first and only family member affected. Genetic counseling is recommended for parents to learn more about Klinefelter syndrome in their family.

How is Klinefelter syndrome treated?

Klinefelter syndrome cannot be cured, but some symptoms can be treated. Infants and toddlers (birth to 3 years) should be connected with *Early On*[®] Michigan if there are concerns about learning, speech, or behavior; while children over 3 years of age should be referred for special education services if concerns arise. Around the time of puberty, testosterone hormone levels should be checked. Many boys have low levels and will benefit from hormone treatment to reduce physical signs of Klinefelter syndrome. Assisted reproduction techniques have helped some men become biological fathers. Children with Klinefelter syndrome and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov



What is Marfan Syndrome?

Marfan syndrome is a genetic condition that occurs in males and females of all racial and ethnic backgrounds. It affects the body's connective tissue and can cause heart, eye, and bone problems. The symptoms vary widely, ranging from mild to severe. Marfan syndrome occurs in about 1 in 5,000 to 1 in 10,000 people.

Michigan Resources & Support

Children's Special Health Care Services

Family Phone Line
Toll-free: 1-800-359-3722
E-mail: ppp@michigan.gov
www.michigan.gov/cshcs

Early On[®] Michigan

Toll-free: 1-800-EARLY ON
www.1800.earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection

www.migeneticsconnection.org

National Resources & Support

National Marfan Foundation

Toll-free: 1-800-862-7326
www.marfan.org

Family Village

www.familyvillage.wisc.edu

GeneReviews

www.geneclinics.org/profiles/marfan/

Genetics Home Reference

www.ghr.nlm.nih.gov/condition=marfan/syndrome

KidsHealth.org

kidshealth.org/kid/health_problems/birth_defect/marfan.html

March of Dimes

www.marchofdimes.com
click on Birth Defects & Genetics

National Institute of Arthritis and Musculoskeletal and Skin Diseases

www.niams.nih.gov/hi/topics/marfan/marfan.htm

How may Marfan syndrome affect my child?

Learning: In general, Marfan syndrome does not affect learning or intelligence. Loose joints may affect large motor skills in some children.

Physical: People with Marfan syndrome are often thin and tall with long limbs relative to the body's torso. Bones and cartilage may be affected, leading to curvature of the spine (scoliosis). The shape of the breast bone may cause a protruding or sunken chest. The joints may be very loose and flexible. The jaw is often narrow with a high-arched palate that can lead to dental crowding.

Medical: Symptoms can range from mild to very severe. Nearsighted vision (myopia) is typical, and often the first symptom identified. A dislocated lens (ectopia lentis) is a common sign often detected only by a special eye exam (slit lamp). Cataracts, glaucoma and retinal detachment may also occur. Heart problems, such as a floppy valve (mitral valve prolapse) are common and can cause shortness of breath and fatigue. The aorta may be wider and more fragile than normal. If not detected, there is a risk of aortic rupture (aneurysm) with serious complications including death.

How does Marfan syndrome occur?

Marfan syndrome is caused by a change in a gene (mutation). A child with Marfan syndrome may be the first and only family member affected, or the gene may be passed down from an affected parent. Parents of a newly diagnosed child should be checked carefully to look for signs of Marfan syndrome. There is a 1 in 2 chance that each child will have Marfan syndrome when a parent carries the gene. Genetic counseling is recommended for parents to learn about possible risks for other family members.

How is Marfan syndrome treated?

Marfan syndrome cannot be cured, but many symptoms can be treated. It is important for a child with Marfan syndrome to be monitored by medical specialists who understand the condition. Most eye problems are corrected with glasses alone but sometimes other procedures are needed. Everyone with Marfan syndrome must be under the care of a heart specialist (cardiologist). Frequent monitoring by ultrasound (echocardiogram) is needed, and medication or surgical procedures may be required. Other therapies or treatments may be needed for health problems as they occur. Infants and toddlers (birth to 3 years) should be connected with *Early On[®]* Michigan if there are concerns about development; while children over 3 years of age should be referred for special education services if concerns arise. Children with Marfan syndrome and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov



What is Neurofibromatosis?

Neurofibromatosis, type 1 (NF1) is a genetic condition that occurs in males and females of all racial and ethnic backgrounds. It mainly changes the development and growth of nerve cells affecting the skin, eyes, nerves and bones of the body. NF1 is found in about 1 in 3,000 people.

Michigan Resources & Support

**Children's Tumor Foundation,
Michigan Chapter**
www.ctf.org/michigan/

NF Support Group of West Michigan
Phone: 616-451-3699
www.nfsupport.org/

**Children's Special Health Care
Services**
Family Phone Line
Toll-free: 1-800-359-3722
E-mail: ppp@michigan.gov
www.michigan.gov/cshcs

Early On[®] Michigan
Toll-free: 1-800-EARLY ON
www.1800earlyon.org

Michigan Birth Defects Program
Nurse Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection
www.migeneticsconnection.org

National Resources & Support

Neurofibromatosis, Inc.
Toll-free: 1-800-942-6825
www.nfinc.org

Family Village
www.familyvillage.wisc.edu

Genetics Home Reference
www.ghr.nlm.nih.gov/ghr/disease/neurofibromatosis/type1

March of Dimes
www.marchofdimes.com, click on Birth Defects & Genetics

**National Institute of Neurological
Disorders and Stroke**
www.ninds.nih.gov/disorders/neurofibromatosis/neurofibromatosis.htm

How may neurofibromatosis affect my child?

Learning: Most people with NF1 have normal intelligence. Learning disabilities are seen in about half the children who have NF1.

Behavior: Attention deficit hyperactivity disorder (ADHD) is common in children with NF1.

Physical: Spots on the skin called "café au lait spots" (meaning the color of coffee with milk) develop during the first few years of life. The spots are present but may not be as obvious in people with NF1 who have darker skin color. Freckles usually develop under the arms or in the groin area. Head size may be larger

than average while a child's height may be shorter than other children the same age. Small nerve tumors called neurofibromas may grow on or under the skin. They are typically benign (non-cancerous). Lisch nodules are tiny bumps that may be seen in the iris of the eyes, but do not affect vision.

Medical: Tumors of the eye nerve (optic gliomas) or other tumors in the brain or spinal cord may develop in childhood. Sometimes cancerous tumors grow along the nerves. There is also an increased risk of other cancers and leukemia. Additional symptoms include bone abnormalities such as curvature of the spine (scoliosis), high blood pressure and seizures.

How does neurofibromatosis occur?

NF1 is caused by a change in a gene (mutation). A child with NF1 may be the first and only family member affected, or the NF1 gene may be passed down from an affected parent. Parents of a newly diagnosed child should be checked carefully to look for signs of NF1. There is a 1 in 2 chance that each child will have NF1 when a parent carries the gene. Genetic counseling is recommended for parents to learn about possible health risks for other family members.

How is neurofibromatosis treated?

NF1 cannot be cured, but some symptoms can be treated. Bone deformities or painful tumors may be treated by surgery. Any tumors that become cancerous may require chemotherapy, radiation or surgery. Yearly eye examinations are recommended to look for signs of optic glioma. Blood pressure should be monitored at regular check-ups. Other treatments may be needed for health problems as they arise. Infants and toddlers (birth to 3 years) should be connected with *Early On[®] Michigan* if there are concerns about learning, speech, or behavior; while children over 3 years of age should be referred for special education services if concerns arise. Children with NF1 and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov

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What is Noonan Syndrome?

Noonan syndrome is a genetic condition that occurs in males and females of all racial and ethnic backgrounds. It can cause certain facial features, heart defects, bleeding problems, short stature, and developmental delays, especially with speech and language. Noonan syndrome occurs in about 1 in 1,000 to 2,500 people.

Michigan Resources & Support

Children's Special Health Care Services

Family Phone Line
Toll-free: 1-800-359-3722
E-mail: ppp@michigan.gov
www.michigan.gov/cshcs

Early On® Michigan

Toll-free: 1-800-EARLY ON
www.1800earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection

www.migeneticsconnection.org

National Resources & Support

Noonan Syndrome Support Group

Toll-free: 1-888-686-2224
www.noonansyndrome.org

Family Village

www.familyvillage.wisc.edu

GeneReviews

www.geneclinics.org/profiles/noonan

Genetics Home Reference

www.ghr.nlm.nih.gov/ghr/disease/noonansyndrome

Human Growth Foundation

Toll-free: 1-800-451-6434
www.hgfound.org

The MAGIC Foundation

Phone: 708-383-0808
www.magicfoundation.org

How may Noonan syndrome affect my child?

Learning: Most children with Noonan syndrome have normal learning. Some may have developmental delays and lifelong learning disabilities. Problems with speech and language are common.

Physical: Facial features may include widely spaced eyes, droopy eyelids (ptosis), and low set ears. These features may be very mild in adults. A broad or webbed neck may be noted. The chest may be an unusual shape with nipples that appear low-set. Many children with

Noonan syndrome are shorter than average.

Medical: Problems with feeding and slow weight gain are common in babies. Between 50 and 80% of children with Noonan syndrome are born with a heart defect. Abnormal bleeding or bruising is common. Mild hearing loss and vision problems can also occur. It is common for males with Noonan syndrome to have undescended testicles (cryptorchidism).

How does Noonan syndrome occur?

Noonan syndrome is caused by a change in a gene (mutation). A child with Noonan syndrome may be the first and only family member affected, or the genetic trait may be passed down from an affected parent. Parents of a newly diagnosed child should be checked carefully to look for signs of the condition. There is a 1 in 2 chance that each child will have Noonan syndrome when a parent carries the gene. Genetic counseling is recommended for parents to learn about the genetic cause of Noonan syndrome in their family, and possible health risks for other children.

How is Noonan syndrome treated?

Noonan syndrome cannot be cured, but many symptoms can be treated. Infants and toddlers (birth to 3 years) should be connected with *Early On*® Michigan if there are concerns about learning, speech, or behavior; while children over 3 years of age should be referred for special education services if concerns arise. Heart defects may be treated with medication or surgery. It is important to check hearing and vision on a regular basis. Lab tests should be done to check blood clotting. Growth should be monitored and use of growth hormone may be considered to increase final adult height. Other treatments may be needed for health problems as they arise. Children with Noonan syndrome and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov



What is Prader-Willi Syndrome?

Prader-Willi syndrome (PWS) is a genetic condition that affects males and females of all racial and ethnic backgrounds. It causes problems with a child's health, behavior and learning. PWS occurs in about 1 in 10,000 to 20,000 people.

Michigan Resources & Support

Prader-Willi Syndrome Association of Michigan

www.pwsausa.org/mi

Children's Special Health Care Services

Family Phone Line

Toll-free: 1-800-359-3722

E-mail: ppp@michigan.gov

www.michigan.gov/cshcs

Early On® Michigan

Toll-free: 1-800-EARLY ON

www.1800earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator

Toll-free: 1-866-852-1247

E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection

www.migeneticsconnection.org

National Resources & Support

Prader-Willi Syndrome Association (USA)

Toll-free: 1-800-926-4797

www.pwsausa.org

Family Village

www.familyvillage.wisc.edu

GeneReviews

www.geneclinics.org/profiles/pws

Genetic and Rare Diseases Information Center

Toll-free: 1-888-205-2311

E-mail: GARDinfo@nih.gov

Genetics Home Reference

www.ghr.nlm.nih.gov/condition=praderwillisynndrome

How may Prader-Willi syndrome affect my child?

Learning: Every child with PWS is unique, but most have developmental and speech delays or learning problems to some degree. The mental impairment is usually mild or moderate. Rarely, a child may have severe mental retardation.

Behavior: Behavioral problems may include sleep disturbance, obsessive-compulsive behavior, temper tantrums and skin picking.

Physical: Children with PWS may have lighter skin, hair and eyes than other family members. They may be short for their age and have smaller hands and feet than average. Small

external genitalia are common. The amount of saliva in the mouth may be less than usual.

Medical: Low muscle tone and trouble with feeding are typical in infancy. Curvature of the spine (scoliosis) and eye muscle problems may be present. Overeating, leading to rapid weight gain, begins in early childhood. Without strict control, obesity, diabetes and other weight-related problems are likely to develop. Low sex hormone levels (hypogonadism) are common, leading to lack of normal physical development and infertility.

How does Prader-Willi syndrome occur?

PWS is caused by changes in genes located on the #15 chromosome. The genetic cause can often be found with special tests. The child is usually the first and only family member affected. Sometimes, the condition can recur in sisters or brothers. Genetic counseling is recommended for parents to learn about the genetic cause of PWS in their family, and possible risks for other children.

How is Prader-Willi syndrome treated?

PWS cannot be cured, but some symptoms can be treated. Special feeding techniques may be needed for a baby with PWS. After the first year or two, food intake will need to be strictly controlled. Behavior therapy may help control the urge to eat. Infants and toddlers (birth to 3 years) should be connected with *Early On*® Michigan as soon as possible. If there are concerns about learning, speech, or behavior in a child over 3 years of age, a referral for special education services should be made. Other therapies or treatments may be needed for developmental or health problems as they arise. Children with PWS and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov.



What is Rett Syndrome?

Rett syndrome is a developmental disorder that affects the brain and nervous system. It occurs mostly in girls of all racial and ethnic backgrounds. After a period of normal development, the condition causes serious developmental problems and difficulty with movement. Rett syndrome occurs in approximately 1 in 10,000 to 15,000 females. It is very rare in males but can occur.

Michigan Resources & Support

Children's Special Health Care Services

Family Phone Line
Toll-free: 1-800-359-3722
E-mail: ppp@michigan.gov
www.michigan.gov/cshcs

Early On® Michigan

Toll-free: 1-800-EARLY ON
www.1800earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection

www.migeneticsconnection.org

National Resources & Support

International Rett Syndrome Association

Toll-free: 1-800-818-7388
www.rettysyndrome.org
Includes RettNet web forum

Rett Syndrome Research Foundation

Phone: 513-874-3020
www.rsrf.org

Family Village

www.familyvillage.wisc.edu

GeneReviews

www.geneclinics.org/profiles/rett

Genetic and Rare Diseases Information Center

Toll-free: 1-888-205-2311
E-mail: GARDinfo@nih.gov

Genetics Home Reference

www.ghr.nlm.nih.gov/condition=rettsyndrome

How may Rett syndrome affect my child?

Learning: Affected girls develop normally until about 6 to 18 months of age. At that time, language and motor skills are quickly lost. Speech and control of hand movements are most often affected. After the time period when skills are lost, the level of development may remain stable. Older children may have slow improvement in some areas.

Behavior: Many girls with Rett syndrome have panic-like attacks and inconsolable crying. Teeth-grinding is a common trait. Some other behaviors may be similar to those found in children with autism.

Physical: Body movement is affected over time. Purposeful hand movements are replaced with repeated wringing, washing, or clapping gestures. Most girls with Rett syndrome are small for their age. Foot and hand deformities and curvature of the spine (scoliosis) are common. Broken bones tend to occur more often than in other children.

Medical: Smaller head size (microcephaly) develops in early childhood. Feeding and digestive problems are common, including constipation. Abnormal breathing patterns and seizures (epilepsy) may also occur.

How does Rett syndrome occur?

Rett syndrome is caused by a change in a gene on the "X" chromosome. The genetic cause can be found in most children with Rett syndrome. The child is usually the first and only family member affected. In rare instances, the condition can recur in sisters or brothers. Genetic counseling is recommended for parents to learn about the genetic cause of Rett syndrome in their family, and possible risks for other children.

How is Rett syndrome treated?

Rett syndrome cannot be cured, but some symptoms can be treated. Infants and toddlers (birth to 3 years) should be connected with *Early On*® Michigan as soon as possible. If there are concerns about learning, speech, or behavior in a child over 3 years of age, a referral for special education services should be made. A speech therapist can help find the best way for a child with Rett syndrome to communicate. Occupational and physical therapy help with movement and mobility. Other therapies or treatments may be needed for problems as they arise. Children with Rett syndrome and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov



What is Tuberous Sclerosis?

Tuberous sclerosis (TS) is a genetic condition that occurs in males and females of all racial and ethnic backgrounds. It often affects the central nervous system, causing seizures, and can lead to non-cancerous tumors in various organs such as the skin, brain, kidneys, heart and eyes. TS occurs in about 1 in 6,000 people.

Michigan Resources & Support

Tuberous Sclerosis Alliance of Michigan

www.tsalliance.org, click on Local Resources

Children's Special Health Care Services

Family Phone Line
Toll-free: 1-800-359-3722
E-mail: ppp@michigan.gov
www.michigan.gov/cshcs

Early On® Michigan

Toll-free: 1-800-EARLY ON
www.1800earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection

www.migeneticsconnection.org

National Resources & Support

Tuberous Sclerosis Alliance

Toll-free: 1-800-225-6872
www.tsalliance.org

Epilepsy Foundation of America

Toll-free: 1-800-332-1000
www.efa.org

Family Village

www.familyvillage.wisc.edu

GeneReviews

www.geneclinics.org/profiles/tuberous-sclerosis

Genetic and Rare Diseases Information Center

Toll-free: 1-888-205-2311
E-mail: GARDinfo@nih.gov

Genetics Home Reference

www.ghr.nlm.nih.gov/ghr/disease/tuberous-sclerosis

How may tuberous sclerosis affect my child?

Learning: Many people with TS have normal intelligence. Others may have lifelong learning problems that range from mild impairment to severe mental retardation.

Behavior: Hyperactivity, attention deficit disorder, or aggressive behavior may occur. Children with TS may also be diagnosed with an autism spectrum disorder.

Physical: Skin changes are common in people with TS. They usually do not lead to serious medical problems, but can cause cosmetic concerns. Skin signs can include white or pale areas, tough and dimpled patches,

facial tumors and fibrous growths around the nails.

Medical: Children with TS often have seizures (epilepsy). The seizures are caused by "tubers", small areas of the brain's outer layer that did not develop normally. Other brain tumors may also occur. Tumors may also form in other organs including the heart, lungs, kidneys and eyes. Some tumors may get smaller and disappear with age while others may grow and cause problems later in life. Most of the tumors are benign (non-cancer) but may have a risk for becoming cancerous.

How does tuberous sclerosis occur?

TS is caused by a change in a gene (mutation). A child with TS may be the first and only family member affected, or the TS gene may be passed down from an affected parent. Parents of a newly diagnosed child should be checked carefully to look for signs of TS. There is a 1 in 2 chance that each child will have TS when a parent carries the gene. Genetic counseling is recommended for parents to learn about possible health risks for other family members.

How is tuberous sclerosis treated?

TS cannot be cured, but some symptoms can be treated. Seizures may be treated by medication. Tumors may be treated by surgery. Imaging studies by ultrasound, CT or MRI should be done to screen for tumors. The frequency and types of studies will depend on symptoms and screening results. Infants and toddlers (birth to 3 years) should be connected with *Early On*® Michigan if there are concerns about learning, speech, or behavior; while children over 3 years of age should be referred for special education services if concerns arise. Other therapies or treatments may be needed for health problems as they occur. Children with TS and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov

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Rev. 4/06



What is Turner Syndrome?

Turner syndrome is a genetic condition that occurs in females of all racial and ethnic backgrounds. It mainly affects growth and fertility, but can also cause other health problems. The condition occurs in about 1 in 2,500 female births.

Michigan Resources & Support

Turner Syndrome Society

- Southeastern Michigan Chapter
- West Michigan Chapter

www.turner-syndrome-us.org

Click on "find a local chapter"

Children's Special Health Care Services

Family Phone Line

Toll-free: 1-800-359-3722

E-mail: ppp@michigan.gov

www.michigan.gov/cshcs

Early On[®] Michigan

Toll-free: 1-800-EARLY ON

www.1800earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator

Toll-free: 1-866-852-1247

E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection

www.migeneticsconnection.org

National Resources & Support

Turner Syndrome Society of the United States

Toll-free: 1-800-365-9944

www.turner-syndrome-us.org

Family Village

www.familyvillage.wisc.edu

Genetics Home Reference

www.ghr.nlm.nih.gov/condition=turnersyndrome

KidsHealth.org

www.kidshealth.org/parent/medical/genetic/turner.html

National Institute of Child Health and Human Development

<http://turners.nichd.nih.gov/>

The MAGIC Foundation

Phone: 708-383-0808

Toll-free Parent Line: 1-800-362-4423

www.magicfoundation.org/www/docs/115/turner_syndrome.html

How may Turner syndrome affect my child?

Learning: In general, girls with Turner syndrome have normal intelligence. They may have learning differences that make verbal learning easier than subjects like math. Some girls have difficulty with visual-spatial skills such as reading maps. Memory and motor coordination may also be affected.

Behavior: Attention Deficit Hyperactivity Disorder (ADHD) sometimes affects behavior in childhood. As girls with Turner syndrome grow up, they may also have concerns with body image and self-esteem.

Physical: One of the most common features is short stature. Girls may also

have other physical features such as lower set ears, droopy eyelids, a low hairline at the back of the neck, a webbed neck and puffy hands/feet.

Medical: Heart defects, kidney problems, and high blood pressure are common. Vision or hearing may be affected. There is a higher chance of diabetes as well as thyroid disorders. Most girls will not begin puberty or have menstrual periods without hormone therapy. Because the ovaries do not develop normally, women with Turner syndrome usually have infertility. Skeletal problems, such as curvature of the spine (scoliosis) and weak bones (osteoporosis) may occur later in life.

How does Turner syndrome occur?

Turner syndrome is caused by a missing "X" chromosome. Usually females have two X chromosomes in all of the body's cells. Girls with Turner syndrome are missing all or part of one X chromosome. A girl with Turner syndrome is usually the first and only family member affected. Genetic counseling is recommended for parents to learn more about Turner syndrome in their family.

How is Turner syndrome treated?

Turner syndrome cannot be cured, but many symptoms can be treated. All girls with Turner syndrome should be checked for heart and kidney defects. Infants and toddlers (birth to 3 years) should be connected with *Early On*[®] Michigan if there are concerns about learning, speech, or behavior; while children over 3 years of age should be referred for special education services if concerns arise. Growth hormone therapy beginning in childhood can help to increase final height as an adult. Estrogen hormone therapy will assist with the onset and progression of normal puberty. Other therapies or treatments may be needed for problems as they arise. Assisted reproductive techniques may allow some women to become pregnant. Girls with Turner syndrome and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based service providers.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov

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Rev. 4/06



What is Williams Syndrome?

Williams syndrome is a genetic condition that occurs in both males and females of all ethnic and racial backgrounds. It affects physical features, health, learning, and behavior. Williams syndrome occurs in about 1 in 7,500 people.

Michigan Resources & Support

Children's Special Health Care Services

Family Phone Line
Toll-free: 1-800-359-3722
E-mail: ppp@michigan.gov
www.michigan.gov/cshcs

Early On® Michigan

Toll-free: 1-800-EARLY ON
www.1800earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection

www.migeneticsconnection.org

National Resources & Support

Williams Syndrome Association

Toll-free: 1-800-806-1871
www.williams-syndrome.org

Williams Syndrome Foundation

Phone: 949-824-7259
www.wsf.org

Family Village

www.familyvillage.wisc.edu

GeneReviews

www.geneclinics.org/profiles/williams

Genetic and Rare Diseases Information Center

Toll-free: 1-888-205-2311
E-mail: GARDinfo@nih.gov

Genetics Home Reference

www.ghr.nlm.nih.gov/condition=williams_syndrome

How may Williams syndrome affect my child?

Learning: Young children often have developmental delays. Older children and adults may have learning disabilities or mild mental impairment. They may also show a great interest or ability in music, and have specific intellectual strengths.

Behavior: Infants may have problems sleeping and be overly sensitive to sound. Children with Williams syndrome often have attention deficit disorder and an "overly-friendly" personality.

Physical: Low birth weight and poor weight gain are common. People with Williams syndrome often have certain

facial features such as a wide mouth, full lips, small teeth, and puffiness around the eyes. The voice may sound hoarse. Soft skin and loose joints are common.

Medical: Feeding problems are common in infants and young children. High calcium levels (hypercalcemia) may occur. This can cause irritability, vomiting, constipation and muscle cramps. Heart and blood vessel disease may develop. A narrowing of the main blood vessel leading from the heart, called supravalvular aortic stenosis, is a frequent finding. Eye problems may also occur.

How does Williams syndrome occur?

Williams syndrome is caused by a change in the #7 chromosome. A very small piece of the chromosome is missing (deleted), including some of the genes within it. The child with Williams syndrome is usually the first and only family member affected. Sometimes the deleted chromosome is passed down from a parent. A person with Williams syndrome has a 1 in 2 (50%) chance of passing the trait on to each of his or her children. Genetic counseling is recommended for parents to learn about the genetic cause of Williams syndrome in their family and possible risks for other children.

How is Williams syndrome treated?

Williams syndrome cannot be cured, but many symptoms can be treated. Infants and toddlers (birth to 3 years) should be connected with *Early On*® Michigan as soon as possible. If there are concerns about learning, speech, or behavior in a child over 3 years of age, a referral for special education services should be made. Calcium levels should be checked, and the child should be monitored for heart problems as well as vision or hearing problems. Heart surgery or other treatments may be needed for health problems as they arise. Children with Williams syndrome and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov